



<110> HEINO, Kai

HEINO, Maarit

PETERSON, Part

SCOTT, Hamish

ANTONARAKIS, Stylianos

LALIOTI, Maria D.

SHIMIZU, Nobuyoshi D.

KUDOH, Jun D.

<120> NOVEL GENE DEFECTIVE IN APECED AND ITS USE

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<140> 09/508,658

<141> 2000-11-03

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<170> PatentIn version 3.2

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Leu His Arg Thr Glu Ile Ala Val Ala Val Asp Ser Ala Phe Pro Leu

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Glu	Thr	Leu	His	Leu	Lys	Glu	Lys	Glu	Gly	Cys	Pro	Gln	Ala	Phe	His		
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Ala	Leu	Leu	Ser	Trp	Leu	Leu	Thr	Gln	Asp	Ser	Thr	Ala	Ile	Leu	Asp		
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Phe	Trp	Arg	Val	Leu	Phe	Lys	Asp	Tyr	Asn	Leu	Glu	Arg	Tyr	Gly	Arg		
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Glu	Asp	Ser	Gly	Ser	Gly	Lys	Asn	Lys	Ala	Arg	Ser	Ser	Ser	Gly	Pro		
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Gly Asp Val Thr Pro Ala Pro Val Glu Gly Val Leu Ala Pro Ser Pro			
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gcc cgc ctg gcc cct ggg cct gcc aag gat gac act gcc agt cac gag			1660
Ala Arg Leu Ala Pro Gly Pro Ala Lys Asp Asp Thr Ala Ser His Glu			
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ccc gct ctg cac agg gat gac ctg gag tcc ctt ctg agc gag cac acc			1708
Pro Ala Leu His Arg Asp Asp Leu Glu Ser Leu Leu Ser Glu His Thr			
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ttc gat ggc atc ctg cag tgg gcc atc cag agc atg gcc cgt ccg gcg			1756
Phe Asp Gly Ile Leu Gln Trp Ala Ile Gln Ser Met Ala Arg Pro Ala			
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Leu Lys Glu Lys Glu Gly Cys Pro Gln Ala Phe His Ala Leu Leu Ser		
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Pro Thr Lys Arg Lys Ala Ser Glu Glu Ala Arg Ala Ala Ala Pro Ala  
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Pro Gln Leu His Gln Lys Asn Glu Asp Glu Cys Ala Val Cys Arg Asp  
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Arg Cys Gly Val Cys Gly Asp Gly Thr Asp Val Leu Arg Cys Thr His  
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Cys Ala Ala Ala Phe His Trp Arg Cys His Phe Pro Ala Gly Thr Ser  
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Pro Gly Pro Ala Lys Asp Asp Thr Ala Ser His Glu Pro Ala Leu His

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Arg Asp Asp Leu Glu Ser Leu Leu Ser Glu His Thr Phe Asp Gly Ile  
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 Met  
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 Trp Leu Val Tyr Ser Ser Gly Ala Pro Gly Thr Gln Gln Pro Ala Arg  
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Val Leu Ala Pro Ser Pro Ala Arg Leu Ala Pro Gly Pro Ala Lys Asp			
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Asp Thr Ala Ser His Glu Pro Ala Leu His Arg Asp Asp Leu Glu Ser			
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Leu Leu Ser Glu His Thr Phe Asp Gly Ile Leu Gln Trp Ala Ile Gln			
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Lys Asn Glu Asp Glu Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile  
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Cys Cys Asp Gly Cys Pro Arg Ala Phe His Leu Ala Cys Leu Ser Pro  
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Gly Glu Glu Val Arg Gly Pro Pro Gly Glu Pro Leu Ala Gly Met Asp  
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Thr Thr Leu Val Tyr Lys His Leu Pro Ala Pro Pro Ser Ala Ala Pro  
195 200 205

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Pro Glu Gly Gln Gln Asn Leu Ala Pro Gly Ala Arg Cys Gly Val Cys  
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Trp Arg Pro Asp Gly Trp Gly Thr Gly Gly Gln Gly Arg Ile Ser Gly  
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Pro Gly Ser Met Gly Ala Gly Gln Arg Leu Gly Ser Ser Gly Thr Gln  
                   50                                  55                                  60

Arg Cys Cys Trp Gly Ser Cys Phe Gly Lys Glu Val Ala Leu Arg Arg  
 65                                  70                                  75                                  80

Val Leu His Pro Ser Pro Val Cys Met Gly Val Ser Cys Leu Cys Gln  
                   85                                  90                                  95

Lys Asn Glu Asp Glu Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile  
                   100                                  105                                  110

Cys Cys Asp Gly Cys Pro Arg Ala Phe His Leu Ala Cys Leu Ser Pro  
115 120 125

Pro Leu Arg Glu Ile Pro Ser Gly Thr Trp Arg Cys Ser Ser Cys Leu  
130 135 140

Gln Ala Thr Val Gln Glu Val Gln Pro Arg Ala Glu Glu Pro Arg Pro  
145 150 155 160

Gln Glu Pro Pro Val Glu Thr Pro Leu Pro Pro Gly Leu Arg Ser Ala  
165 170 175

Gly Glu Glu Pro Arg Cys Gln Gly Trp Thr Pro Arg Pro Cys Thr Pro  
180 185 190

Tyr Cys Val Trp Val Leu Arg Val Ser Arg Thr Trp Leu Leu Val Arg  
195 200 205

Val Ala Gly Cys Ala Glu Met Val Arg Thr Cys Cys Gly Val Leu Thr  
210 215 220

Ala Pro Leu Pro Ser Thr Gly Ala Ala Thr Ser Gln Pro Ala Pro Pro  
225 230 235 240

Gly Pro Gly Arg Ala Cys Ala Ala Asp Pro Ala Gln Glu Thr  
245 250

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20

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<223> Description of Artificial Sequence: PRIMER

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22

<210> 18

<211> 22

<212> DNA

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<400> 18

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22

<210> 19

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21

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acgggctcct caaacaccac t

21

<210> 21

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<212> DNA

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<400> 21

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24

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24

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24

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1 5 10 15

<210> 26  
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<400> 33  
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<400> 34  
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<400> 35

ggactgagga aggaggtgtc cttc

24

<210> 36

<211> 20

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<223> Description of Artificial Sequence: PRIMER

<400> 36

Asp Gly Ile Leu Gln Trp Ala Ile Gln Ser Met Ala Arg Pro Ala Ala  
1 5 10 15

Pro Phe Pro Ser  
20

<210> 37

<211> 42

<212> PRT

<213> Homo sapiens

<400> 37

Cys Ala Val Cys Arg Asp Gly Gly Glu Leu Ile Cys Cys Asp Gly Cys  
1 5 10 15

Pro Arg Ala Phe His Leu Ala Cys Leu Ser Pro Pro Leu Arg Glu Ile  
20 25 30

Pro Ser Gly Thr Trp Arg Cys Ser Ser Cys  
35 40

<210> 38

<211> 42

<212> PRT

<213> HOMO SAPIENS

<400> 38

Cys Gly Val Cys Gly Asp Gly Thr Asp Val Leu Arg Cys Thr His Cys  
1 5 10 15

Ala Ala Ala Phe His Trp Arg Cys His Phe Pro Ala Gly Thr Ser Arg  
20 25 30

Pro Gly Thr Gly Leu Arg Cys Arg Ser Cys  
35 40

<210> 39  
<211> 42  
<212> PRT  
<213> HOMO SAPIENS

<400> 39

Cys Glu Val Cys Gln Gln Gly Gly Glu Ile Ile Leu Cys Asp Thr Cys  
1 5 10 15

Pro Arg Ala Thr His Met Val Cys Leu Asp Pro Asp Met Glu Lys Ala  
20 25 30

Pro Glu Gly Leu Trp Ser Cys Pro His Cys  
35 40

<210> 40  
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<212> PRT  
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<400> 40

Cys Arg Val Cys Lys Asp Gly Gly Glu Leu Ile Cys Cys Asp Thr Cys  
1 5 10 15

Pro Ser Ser Tyr His Ile His Cys Leu Asn Pro Pro Leu Pro Glu Ile  
20 25 30

Pro Asn Gly Glu Trp Leu Cys Pro Arg Cys  
35 40

<210> 41  
<211> 42  
<212> PRT  
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<400> 41

Cys Ala Val Cys Gln Asn Gly Gly Glu Leu Ile Cys Cys Glu Lys Cys  
1 5 10 15

Pro Lys Val Phe His Leu Ser Cys His Val Pro Thr Leu Thr Asn Phe  
20 25 30

Pro Ser Gly Glu Trp Ile Cys Thr Phe Cys  
35 40